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Students Online: Learning Medical Genetics

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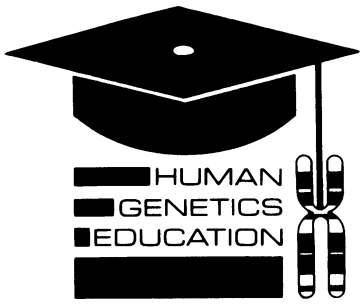

Summary

It is possible to focus medical genetics education by using a model that integrates the skills of end-user searching of the medical literature into the traditional course content. Since 1988, 313 first-year medical students were studied as they accessed MEDLINE to retrieve information about biochemical genetic disorders. Their search behavior was studied by analyzing data from the National Library of Medicine's traffic files. The skills that they initially learned were reinforced as they searched clinical genetics problem cases in the second-year pathology course, and these skills were consolidated in the third year when the students addressed specific patient-care questions in pediatrics. The students' perception of the value of this model was studied by analyzing questionnaires completed during the exercise. It was demonstrated that when students were taught the skills of accessing MEDLINE by computer, they could formulate a question, retrieve current information, critically review relevant articles, communicate effectively, and use these skills to contribute to patient care.

Introduction

Many medical schools are instituting major curricular changes using computers (Ball and Douglas 1989; Friedman et al. 1990*a*, 1990*b*; Lynch and Jaffe 1990). Strategies, such as using problem-based examinations to assess clinical case management, attempt to measure clinical acumen rather than factual recall (Stevens et al. 1989; Rubeck 1990). Students are learning how to access and analyze a massive, dynamic data base of medical information. The rapid proliferation of literature in human genetics and the critical nature of molecular applications for diagnosis and treatment make education in medical genetics ideal for innovative strategies.

In the study described here, the primary goal was to teach the skills of using computers to search the literature. Exercises to teach concepts of medical genetics were incorporated in courses in biochemistry and pathology. While practicing these skills, the students also learned how to define a problem in medical genetics, formulate a question, retrieve recent information, review relevant articles, and communicate the information learned. Secondary goals included expanding the teaching of medical genetics and integrating clinical and basic science education into the medical curriculum.

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Material and Methods

Genetics in Biochemistry

In 1988, 1989, and 1990, first-year biochemistry classes were taught to search MEDLINE by using Grateful Med (National Library of Medicine 1991). Each student was required to update information about a specific disorder listed in the *Catalogs of Mendelian Inheritance in Man* (*MIM*) (McKusick 1988, 1990), to write a one-page abstract of new information, and to present this information in a small group discussion session led by both a geneticist and a biochemist, following a format originally described by Bodurtha et al. (1986). Two supplementary computer software

packages were available as resources for additional study of the basic principles of human genetics. Each student was encouraged to use his or her MEDLINE access code to search independently during the 5-wk exercise. The student-search sessions were analyzed by study of the National Library of Medicine (NLM) traffic files, with a signed release. Detailed description of the procedures for analysis can be found in the report by Mitchell et al. (1992). Student attitudes were evaluated when the students completed a questionnaire at the end of each discussion session, as described in previous reports (Proud et al. 1989; Mitchell et al. 1990).

The computer laboratories and discussion sessions were staffed by a geneticist and a biochemist. Librarians also participated in the computer laboratories. Topics were grouped in order to focus the search questions. For example, in the session on "Hematology and Blood Disorders," each student searched on a topic such as hemophilia (MIM 306700), sickle cell disease (MIM 142300), adenosine deaminase deficiency (MIM 102700), or chronic granulomatous disease (MIM 306400). Another laboratory and discussion group addressed questions on "Skeletal and Connective Tissue Problems," such as osteogenesis imperfecta (MIM 259400), homocystinuria (MIM 236200), Menkes syndrome (MIM 309400), and steroid sulfatase deficiency (MIM 308100). Other sessions addressed topics in "Amino Acid Metabolism," "Lipid Metabolism," "Renal and Endocrine Disorders," "Carbohydrate Storage Disorders," and "Neurologic Disorders." Concepts about autosomal dominant, autosomal recessive, and X-linked conditions were addressed in each session. The students searched a subtopic that emphasized basic principles in genetics, such as molecular mechanisms, inheritance patterns, linkage, chromosome mapping, prenatal diagnosis, newborn screening, or therapy. They used medical subject headings (MeSH) to complete the search screen, ran the searches, refocused as necessary, and retrieved up to five references for review. They prepared a brief abstract of recent information and presented their findings in the discussion session.

Several types of data were analyzed. First, search strategies and skills were extracted from in-depth review of the traffic files obtained from the NLM and were identified by individual student passwords. Errors in search techniques were coded as Boolean logic errors, redundant searches, incorrect use of terminology, misspelling, or other incorrect use of the software. Second, student attitudes were analyzed from questionnaires completed at the end of the discussion sessions. Virtually all students who were asked completed the questionnaires. Third, validity of the material and student comprehension were determined from written and oral reports. Statistical analyses were performed by using the Kruskal-Wallis test for χ^2 approximation and by comparing group means between different groups as they participated in each aspect of the study. Paired data were used when the results were analyzed longitudinally over the 3 years.

Genetics in Pathology

The primary goal of this part of the study was to reinforce the computer expertise acquired in the biochemistry course, by means of a second opportunity to perform MEDLINE searches. Prior to lectures on chromosomes, Mendelian inheritance, and multifactorial inheritance and teratogens, the students were given questions about a clinical case, such as an infant with trisomy 18, a child with muscular dystrophy, or a toddler with fetal alcohol syndrome. The students worked in groups of four to access MEDLINE and to retrieve relevant articles. A clinical geneticist or genetics counselor and a librarian staffed the computer laboratories. The focus of each computer laboratory session moved from simply teaching literature-searching skills to encouraging more difficult searches that required the students to acquire some knowledge about the topics before entering MEDLINE. The students needed to read about basic clinical genetics by using the available texts and reference materials, such as *Robbin's Pathologic Basis of Disease* (Cotran et al. 1989) and *Genetics in Medicine* by Thompson and Thompson (1986), in order to come to the computer laboratory prepared to structure the search and to retrieve relevant articles. The students then led the discussion sessions.

Literature Searching in Pediatrics

Beginning in July 1989, each student in the third-year pediatric clerkship was required to search at least one topic relating to patient care. The aim of this exercise was to provide another opportunity to formulate a question and to solve a problem, while consolidating skills in computer literature searching. These activities were primarily monitored by the attending physician. Students were required to complete a written report and to present their findings, in a seminar format at the end of the clerkship, to the clinical geneticist and a librarian. The content and quality of the presentations were evaluated.

Longitudinal Study

Sixteen randomly chosen students were restudied, and their search data were analyzed longitudinally over

Table 1

Traffic-File Data from Student Searches Performed in the Genetics in Biochemistry Exercise in the First 2 Years

	1988	1989	*P* value[a]
Searches	825	590	...
Searches/student	7.24	6.63	N.S.
Students	114	105	...
Students doing independent searches	91	89	...
Genetics searches	299	476	...
Students searching	50	89	...
Searches per student	5.98	5.35	N.S.
Nongenetics searches	526	114	...
Students searching	78	21	...
Searches per student	6.74	5.43	N.S.
Searches with errors	11%	5%	<.01

[a] N.S. = not significant.

the 3-year experience in biochemistry, pathology, and pediatrics. Telephone interview data and traffic files were analyzed in order to determine whether there was improvement in the students' ability to search during the 3 years. The purpose was to identify factors that could determine which students were likely to effectively use, in patient-care situations, this kind of literature searching by computer.

Results

Genetics in Biochemistry

Three hundred thirteen students used MEDLINE codes to perform more than 2,000 independent searches of the medical literature during the 3-year period. Hundreds of additional searches were also completed during the computer training labs. Data analysis from the first 2 years is detailed in table 1. Students performed an average of 6.9 independent searches over the first 2 years. At student rates from the NLM, the cost for the independent searches would average approximately $10.00/student. The percentage of searches with errors decreased from 11% in 1988 to 5% in 1989 ($P < .01$). Analysis of the 1989 data revealed that students who did more searches had more searches with errors ($r = .285$; $P < .01$), which is consistent with studies by Bresnitz et al. (1986). This is in contrast to our 1988 data analysis (Proud et al. 1989), which suggested that students who searched more were better and had fewer errors. The 10 students who reported owning a computer also did more searches than the average but had fewer searches with errors. Data on these 10 students were inadequate for performing statistical analysis, because of the small numbers. Fewer total independent searches in 1989 may reflect several factors, including fewer students, grouping of topics in the computer laboratories and discussion sessions, timing of spring vacation, and unique class differences in attitude and approach.

Consistently in each year, 97%–98% of the students completing the questionnaire reported that the experience was a "satisfactory" or "excellent" way to learn literature searching by computer (table 2). Seventy percent of the students reported that it was a "satisfactory" or "excellent" way to learn basic concepts in genetics. Conversely, 30% of students noted that they did not feel this was a "satisfactory" way to learn genetics. It was not possible to determine whether these were also the students who did few independent searches. Student criticism of the discussion sessions declined from 20% to 7% "unsatisfactory," while approval as "good" or "excellent" increased from 80% to 93% when results between 1988 and 1990 are compared.

Genetics in Pathology

Comparison of the traffic-file data of students who had biochemistry, in March 1988, followed by pathology, in September 1988, revealed differences in the student search frequency (fig. 1). Individuals who did no independent searches in the first year were sometimes frequent searchers 6 mo later. Analysis of the change in error rate revealed that students who made errors in biochemistry demonstrated a significant decrease in error rate in pathology. Twenty percent of the biochemistry searches had at least one error, while only 11% of the pathology searches had at least one error ($P < .01$), suggesting improved searching performance.

Another factor that suggests that the implementation of this exercise in pathology may have improved learning was the consistent observation, by the discussion leaders, of the enthusiasm with which the students taught interesting aspects of their clinical cases to their peers. The discussion leaders acted as facilitators who clarified concepts, pointed out errors in content, and guided the students to use their newly acquired knowledge to understand basic concepts in clinical genetics and pathology.

Literature Searching in Pediatrics

Two cases illustrate the way in which student literature searching had an impact on clinical patient care:

Case 1.—An 8-mo-old male with G-syndrome (dysmorphic face with hypertelorism, hypotonia, hypospa-

Table 2

Genetics in Biochemistry: Value of the Experience

Session or Activity	% of Student Responses to Questionnaires							
	1988 (n = 91)				1990 (n = 82)			
	Unsatisfactory	Satisfactory	Excellent	Combined Satisfactory and Excellent	Unsatisfactory	Satisfactory	Excellent	Combined Satisfactory and Excellent
Discussion session	19.8	67.0	13.2	80.2	7.4*	65.4	27.2	92.6
Genetics principles	33.1	61.1	7.8	68.9	28.4	56.8	14.8	71.6
Biochemistry	24.4	70.0	5.6	75.6	22.3	56.4	21.3**	77.7
Computer searching	1.1	41.8	57.1	98.9	2.5	53.1	44.4	97.5

NOTE.—χ^2 comparisons were made between category means for each year, 1988 and 1990.
* $P < .01$.
** $P < .001$.

dias, and developmental delay) presented with respiratory distress and tracheomalacia (Winter and Baraitser 1990). Tracheostomy was performed, but cor pulmonale persisted. Attempts to wean him to room air resulted in frequent episodes of apnea and bradycardia. Digoxin treatment was considered. The third-year student used Grateful Med to access MEDLINE in order to retrieve references containing the latest recommendations about treatment of infants with cor pulmonale. He confirmed the inefficacy of digoxin treatment, and his review of the pathophysiology of cor pulmonale suggested that chronic hypoxia was probably contributing to the apnea and exacerbating the right-heart failure. With this information, it was decided to maintain the ambient oxygen concentration at 40%. Within 36 h, the child demonstrated no further episodes of apnea. Repeat echocardiograms documented resolution of the cor pulmonale. The child was discharged, with supplemental oxygen. By modifying the treatment plan on the basis of both a review of the basic science and information retrieved from the literature, it was possible to decrease patient time in the hospital and potentially improve his overall prognosis.

Case 2.—A newborn female presented with an unusual skin rash. Initial dermatologic evaluation suggested linear sebaceous nevus syndrome (MIM 163200). The medical student reviewed the literature and identified a physician who had authored a number of publications on this topic. The family elected to transport the infant over 500 miles for further evaluation by the expert identified through the literature search. A final diagnosis of chondrodysplasia punctata (MIM 255100) was made. Clearly, the information gained from the literature review significantly altered the evaluation and management of this child.

Longitudinal Study

Data from interview questions and traffic files of 16 randomly chosen students were analyzed by comparing group means in biochemistry, pathology, and pediatrics. Fifty-six percent of the students felt insecure about their searching ability and would have preferred to have a librarian available to assist in the patient-care searches. Ninety-four percent again reported that their experience with Grateful Med was "satisfactory" or "excellent." Individual search quality was studied by reviewing error rates. Twenty-five percent of the students overestimated their ability to use the software "effectively." Further studies will be needed to determine which students are effective searchers. Retrieving 20 articles but missing a key review may have serious clinical implications.

Discussion

Computer applications, including computer-assisted instruction, patient simulations, examinations, and bibliographic searches, are rapidly becoming the rule in medical education (Friedman et al. 1990 *a*, 1990*b*; Mitchell 1991). This study describes a simple exercise that uses a computer laboratory and that provides indepen-

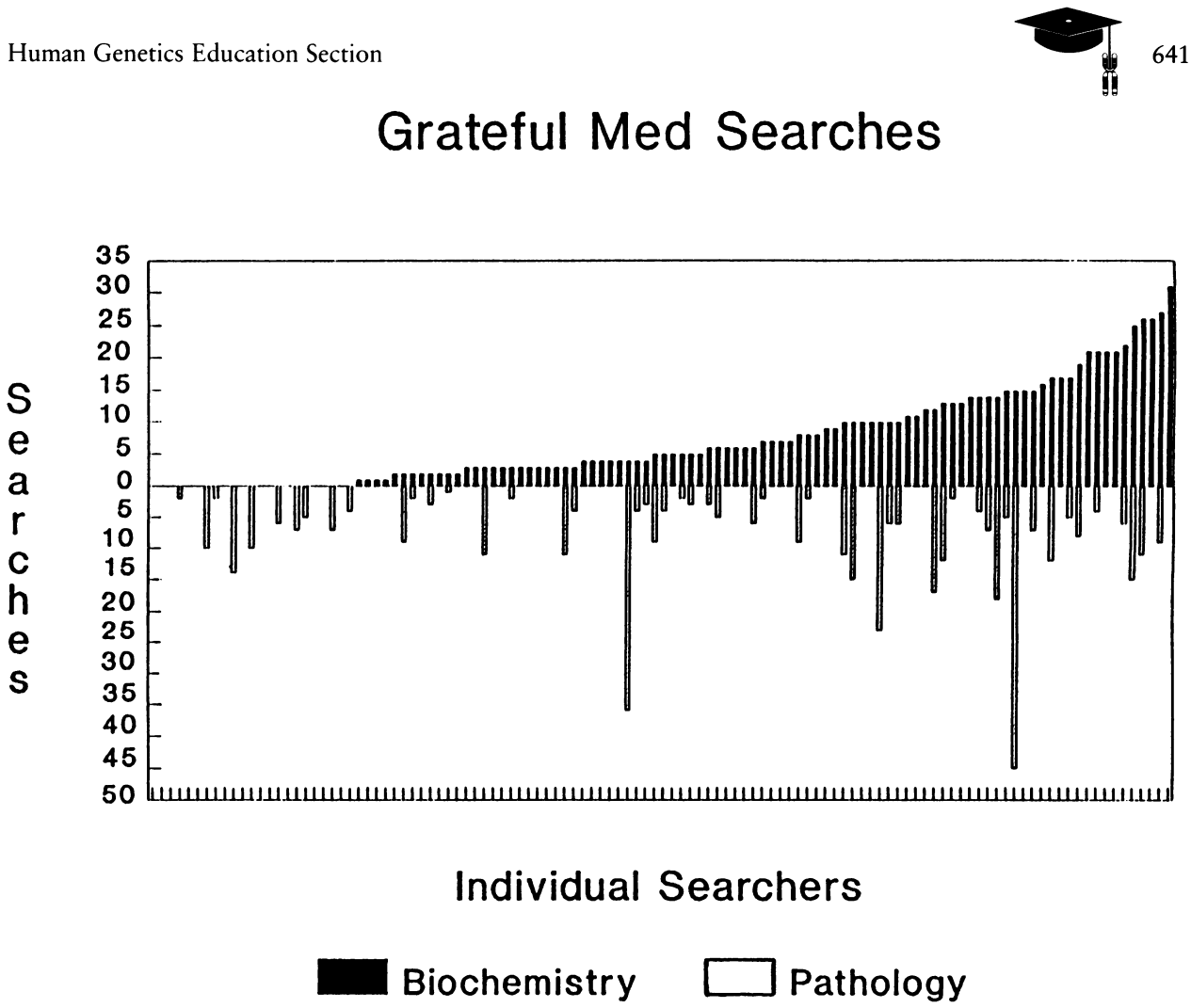

Figure 1 No. of independent searches done by each of 115 students during the 1989 genetics in biochemistry exercise followed by the pathology exercise. (Reprinted with permission from Academic Press, 1992)

dent search time to teach the skills of using a computer for literature access early in the medical career, in order to integrate information about medical genetics into the curriculum. Teaching online bibliographic search strategies and skills can be a model for teaching the use of computers to access a variety of data bases.

This study was begun at a time when the students received no formal instruction in computer use. The only control group was found in a retrospective review of student experience using computers before institution of these exercises. Obviously, this limits the interpretation of the effectiveness of this model. However, it is clear that during this 3-year period hundreds of students learned the skills necessary to access MEDLINE by using Grateful Med, performed thousands of searches, and became aware of the easy accessibility to the vast current literature in medical genetics. Other investigators have reported a curve for users of Grateful Med, which indicated that a small number of individuals did the majority of the searches (Haynes et al. 1990). In contrast, the majority of the students reported here used Grateful Med to access MEDLINE in both basic science courses and clinical rotation.

Several problems have been identified. This kind of exercise cannot substitute for basic instruction in the principles of genetics. Improved emphasis on software review and competency testing must be incorporated into future studies. The students should use the computers only after they understand core concepts and vocabulary in genetics. They must be able to pose an appropriate question in order to structure an effective search. Grateful Med can help the student generate appropriate vocabulary by using MeSH in order to define the scope of the literature about a specific topic. Often,

by running several searches, a student can gain enough information about the topic to structure a search that results in relevant references.

The students must learn that reading abstracts from MEDLINE can never substitute for critical review of journal articles. Likewise, journal articles cannot substitute for basic medical facts. When applying these search skills to patient-care problems, the student must learn to integrate information from the literature with data from the history and physical exam. Information from the literature must be cautiously interpreted when applied in clinical practice. It will be important to develop objective measures of the application of these skills and of the impact that this knowledge has in the clinical years. Currently, studies are under way to improve this model. In order to measure the effectiveness of student searches, it will be necessary to determine whether the students are actually retrieving key or essential articles. This is being done by having the librarian generate lists of appropriate references by using MEDLINE together with other data bases, such as the Online Mendelian Inheritance in Man (Genome Data Base/OMIM 1992).

Innovative education in medical genetics will provide opportunities for collaboration between basic scientists and clinicians. There are many potential applications of this model. It should be possible to improve genetics education by applying the skills of computer use and bibliographic searching to many other areas of medical education.

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